

10/527831

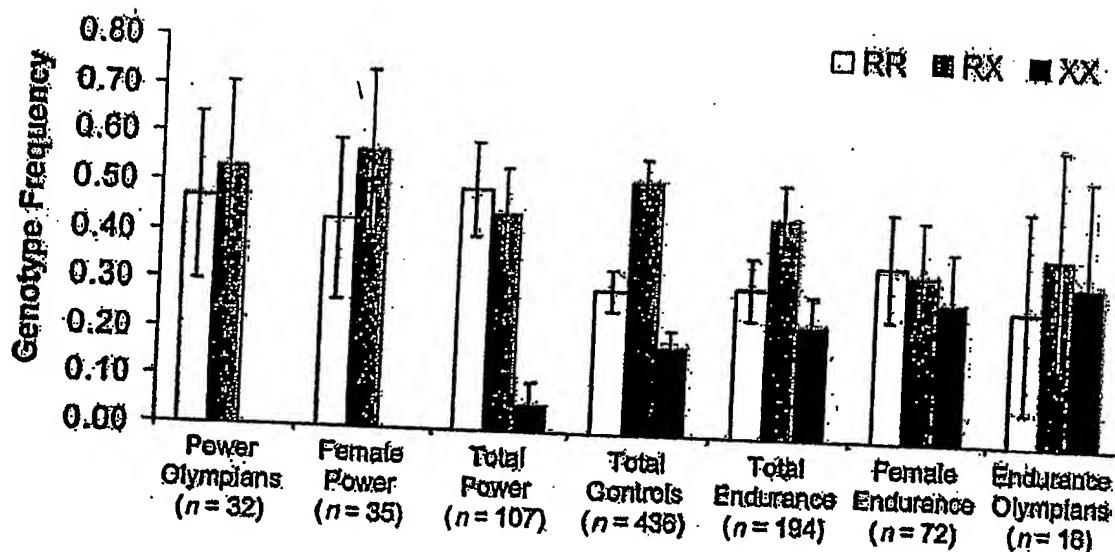


Fig. 1

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Table 1

Number and Frequency (%) of *ACTN3* Genotypes and Frequency (%) of *ACTN3* Alleles in Controls and Elite Sprint/Power and Endurance Athletes

GROUP (n)	No. (%) WITH GENOTYPE			ALLEL FREQUENCY (%)	
	RR	RX	XX	R	X
Male:					
Controls (134)	40 (30)	73 (54)	21 (16)	57	43
Sprint (72)	38 (53)	28 (39)	6 (8)	72	28
Endurance (122)	34 (28)	63 (52)	25 (20)	54	46
Female:					
Controls (292)	88 (30)	147 (50)	57 (20)	55	45
Sprint (35)	15 (43)	20 (57)	0 (0)	71	29
Endurance (72)	26 (36)	25 (35)	21 (29)	53	47
Total:					
Controls (436)	130 (30)	226 (52)	80 (18)	56	44
Sprint (107)	53 (50)	48 (45)	6 (6)	72	28
Endurance (194)	60 (31)	88 (45)	46 (24)	54	46

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TABLE 2: Genotyping of R577X in ACTN3 in Caucasians Elite Athletes.

Strength	Sport	ID	Sport Institute	Total Number	577RR (%)	577RX (%)	577XX (%)
Endurance	Rower	RT492	AIS	64	22 (34.4%)	28 (43.8%)	14 (21.8%)
		RT556					
Endurance	Triathloner	RT977	AIS	13	3 (23.1%)	8 (61.5%)	2 (15.4%)
		RT989					
Endurance	Cyclist	RT990	AIS	9	4 (44.4%)	2 (22.2%)	3 (33.3%)
		RT998					
Endurance	Track Cyclist	KN246	AIS	22	7 (31.8%)	7 (31.8%)	8 (36.4%)
		KN275					
Endurance	Marathon	KN310	AIS	1	0	0	1
Endurance	All above		AIS	108	36 (33.3%)	45 (41.7%)	27 (25.0%)
Sprint	Swimmer	RT901	AIS	45	17 (37.8%)	25 (55.6%)	3 (6.6%)
		RT1018					
Sprint	Track Cyclist	KN246	AIS	8	4 (50.0%)	3 (37.5%)	1 (12.5%)
		KN275					
Sprint	Athletics	KN276	AIS	30	16 (53.3%)	13 (43.3%)	1 (3.3%)
		KN309					
Sprint	All above		AIS	83	37 (44.6%)	41 (49.4%)	5 (6.0%)
Africa Zulu				88	69 (78.4%)	18 (20.5%)	1 (1.1%)
Australian Caucasian Control				152	46 (30.0%)	78 (52.0%)	28 (18%)

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Table 3 SNPs identified in the ACTN3 gene to date
NCBI SNP CLUSTER ID

rs2229456

rs2229455

rs2229454

rs2228325

rs1126675

rs7949754

rs7924602

rs5792393

rs4990284

rs4990283

rs4013815

rs3937320

rs3837428

rs3814736

rs3814735

rs3782080

rs2511217

rs2511216

rs2509559

rs2509558

rs2305537

rs2305534

rs2290463

rs2275998

rs2096583

rs2000939

rs1815739

rs1791690

rs1671064

10/527831

rs1188610

rs679228

rs678397

rs677488

rs647476

rs647029

rs618838

rs607736

rs597626

rs544021

rs540874

rs538330

rs531490

rs509556

rs490998

rs13897

rs4576

rs1189338

rs1201433

rs640213

rs3737525

rs3178740

rs3180065

rs3180064

rs3180063

rs3867132

rs608504

rs610293

rs3825065

www.ncbi.nlm.nih.gov/SNP/snp_ref

TSC: The SNP Consortium website

TABLE 4. Symbols, full names, and cytogenic location of nuclear and mitochondrial genes of the 2002 Human Gene Map for Performance and Health-Related Fitness Phenotypes.

Gene or Locus Name Location

A B

ACADVL Acyl coenzyme A dehydrogenase, very long chain 17p13-p11

ACE Angiotensin I converting enzyme 17q23

ADRA2A Alpha-2A-adrenergic receptor 10q24-q26

ADRB1 Adrenergic, beta-1-, receptor 10q24-q26

ADRB2 Beta-2-adrenergic receptor 5q31-q32

ADRB3 Beta-3-adrenergic receptor 8p12-p11.2

AGT Angiotensinogen 1q42-q43

ANG Angiogenin, ribonuclease, RNase A family, 5 14q11.1-q11.2

APOE Apolipoprotein E 19q13.2

ATP1A2 ATPase, Na₊/K₋ transporting, alpha-2 polypeptide 1q21-q23

ATP1B1 ATPase, Na₊/K₋ transporting, beta 1 polypeptide 1q22-q25

BDKRB2 Bradykinin receptor B2 14q32.1-q32.2

C D E F G

CASQ2 Calsequestrin 2 (cardiac muscle) 1p13.3-p11

CFTR Cystic fibrosis transmembrane conductance regulator, ATP-binding cassette (subfamily C, member 7) 7q31.2

CKM Creatine kinase, muscle 19q13.2-q13.3

CNTF Ciliary neurotrophic factor 11q12.2

CPT2 Carnitine palmitoyltransferase 2 1p32

COL1A1 Collagen, type I, alpha 1 17q21.3-q22.1

EDN1 Endothelin 1 6p24.1

ENO3 Enolase 3, (beta, muscle) 17pter-p11

FABP2 Fatty acid binding protein 2 4q28-q31

FGA Fibrinogen, A alpha polypeptide 4q28

FGB Fibrinogen, B beta polypeptide 4q28

GDF8 (MSTN) Growth differentiation factor 8 (myostatin) 2q32.2

GNB3 Guanine nucleotide binding protein (G protein), beta polypeptide 3 12p13

H I K L M

HLA-A Major histocompatibility complex, class I, A 6p21.3

HP Haptoglobin 16q22.1

IGF1 Insulin-like growth factor I 12q22-q23

IGF2 Insulin-like growth factor 2 11p15.5

IL-6 Interleukin-6

KCNQ1 K₊voltage-gated channel, KQT-like subfamily, member 1 11p15.5

LDHA Lactate dehydrogenase A 11p15.4

LPL Lipoprotein lipase 8p22

MTCO1 Cytochrome c oxidase I mtDNA 5904-7445

MTCO3 Cytochrome c oxidase III mtDNA 9207-9990

MTCYB Cytochrome b mtDNA 14747-15887

MTND1 NADH dehydrogenase 1 mtDNA 3307-4262

MTND4 NADH dehydrogenase 4 mtDNA 10760-12137

MTND5 NADH dehydrogenase 5 mtDNA 12337-14148
MTTE Transfer RNA, mitochondrial, glutamic acid mtDNA 14674-14742
MTTI Transfer RNA, mitochondrial, isoleucine mtDNA 4263-4331
MTTK Transfer RNA, mitochondrial, lysine mtDNA 8295-8364
MTTL1 Transfer RNA, mitochondrial, leucine 1 (UUR) mtDNA 3230-3304
MTTL2 Transfer RNA, mitochondrial, leucine 2 (CUN) mtDNA 12266-12336
MTTM Transfer RNA, mitochondrial, methionine mtDNA 4402-4469
MTTT Transfer RNA, mitochondrial, threonine mtDNA 15888-15953
MTTY Transfer RNA, mitochondrial, tyrosine mtDNA 5826-5891
MyHC myosin Heavy-chain
N O P Q R S T U V
NOS3 Nitric oxide synthase 3 (endothelial cell) 7q36
NPY Neuropeptide Y 7p15.1
PAI1 Plasminogen activator inhibitor 1 7q21.3-q22
PFKM Phosphofructokinase, muscle 12q13.3
PGAM2 Phosphoglycerate mutase 2 (muscle) 7p13-p12
PGK1 Phosphoglycerate kinase 1 Xq13
PHKA1 Phosphorylase kinase, alpha 1 (muscle) Xq12-q13
PON1 Paraoxonase 1 7q21.3
PPARA Peroxisome proliferative activated receptor, alpha 22q13.31
PPARG Peroxisome proliferative activated receptor, gamma 3p25
PYGM Phosphorylase, glycogen, muscle 11q12-q13.2
RYR2 Ryanodine receptor 2 (cardiac) 1q42.1-q43
SGCA Sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein) 17q21
S100A1 S100 calcium binding protein A1 1q21
SUR Sulfonylurea receptor 11p15.1
TGFB1 Transforming growth factor beta 1 19q13.2
UCP2 Uncoupling protein 2 11q13
UCP3 Uncoupling protein 3 11q13
VDR Vitamin D (1,25-dihydroxyvitamin D3) receptor 12q12-q14
The gene symbols, names and cytogenetic locations are from the Locus Link web site
(<http://www.ncbi.nlm.nih.gov/LocusLink>) available from the National Center for
Biotechnology
Information (NCBI). For mitochondrial DNA, locations are from the human
mitochondrial genome data base (<http://www.mitomap.org>).

TABLE 5. Endurance phenotypes and case-control studies (DNA polymorphisms).

Gene	Location	Athletes			Controls		
		N	Sports	Freq.	N	Freq.	P
<i>ADRA2A</i>	10q24-q26	140	Endurance	6.7/6.7: 0.77	141	6.7/6.7: 0.62	0.037
				6.7/6.3: 0.21		6.7/6.3: 0.34	
				6.3/6.3: 0.02		6.3/6.3: 0.04	
				6.7: 0.88		6.7: 0.8	0.011
				6.3: 0.12		6.3: 0.2	
<i>ACE</i>	17q23	64	Endurance	II: 0.30	118	II: 0.18	0.03
				ID: 0.55		ID: 0.51	
				DD: 0.16		DD: 0.32	
				I: 0.57		I: 0.43	0.02
				D: 0.43		D: 0.57	
	79	Running		I: 0.57	Ref. Pop. Ref.	I: 0.49	0.039
				D: 0.43		D: 0.51	
				NA		NA	
				II: 0.25		Pop. Ref.	0.003
	25	Mountain-eering			Ref. Pop. Ref.	II: 0.16	0.0009
	60	Elite athelets (cycling, running, handball)		ID: 0.58	Pop. Ref.	ID: 0.45	
				DD: 0.17		DD: 0.39	
				I: 0.54		I: 0.38	
				D: 0.46		D: 0.62	
				II: 0.15		1248 II: 0.24	0.004
	56	Elite swimmers (subsample of 103 swimmers)		ID: 0.39	Pop. Ref.	ID: 0.49	
				DD: 0.46		DD: 0.27	
				I: 0.34		I: 0.48	
				D: 0.66		D: 0.52	

Reference: Perusse et al. 2003 "The human gene map for performance and health-related fitness phenotypes: the 2002 update" Med. Sci. Sports Exerc. 35: 1248-1264.

TABLE 6. Genotype and allelic frequencies of *ACTN3* 577R/X alleles in human populations.

Ethnic group	No. of chromosomes	No. of genotypes		Relative allele frequency of 577X
		RX	XX	
Asian	56	14	7	0.5+0.07
Javanese	96	28	12	0.54+0.05
Native American	14	2	2	0.43+0.14
Asia/Americas	166	44	21	0.52+0.04
Hispanic	64	16	5	0.41+0.06
White	214	47	21	0.42+0.03
Europe	278	63	26	0.41+0.03
Aboriginal	174	33	9	0.29+0.03
Australian				
PNG Highlander	78	16	6	0.36+0.05
Australasia	252	49	15	0.31+0.03
African American	90	12	6	0.27+0.05
African Bantu	156	14	1	0.10+0.05
Africa	246	56	7	0.16+0.05
Unknown	152	50	11	0.47
Total	1094	232	80	